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A Rare Case of Pierre Robin Sequence

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Abstract: ***PIERRE ROBIN SEQUENCE is a set of abnormalities affecting the head and face, consisting of a small lower jaw (micrognathia), a tongue that is placed further back than normal (glossoptosis), and blockage of the airways. We report a male neonate baby delivered through normal vaginal delivery and having features of PIERRE ROBIN SEQUENCE.***

Keywords: Pierre Robin Sequence, Micrognathia, Glossoptosis.

1. Introduction

Pierre robin sequence is clasically defined as micrognathia, glossoptosis, and airway obstruction.cleft palate is found in roughly 90% of patients. [1]. It is an autosomal recessive disorder. The prevalence ranging from 1:20,000 to1:30,000 live births[1]. The most concerning complications are respiratory distress, poor feeding, speech difficulties, aspiration and gastroesophageal reflux due to micrognathia, cleft palate and repostional tongue.

2. Case Report

A Day 1 old male neonte born to a 2 degree consanguinous couple by normal vaginal delivery presented with perinatal asphyxia HIE stage two and respiratory distress.

On examination child had micrognathia, retrognathia, cleft of soft palate, inspiratory stridor and lower chest and upper chest retractions.

His Vitals were Heart rate of 130/min, Respiratory rate of 82/min, SPO2- 82% @Room Air.

Biochemical investigations are normal. Sepsis screen is positive, PT, APTT prolonged, Liver functions tests were normal and other investigations were found to be normal.

On direct laryngoscopy: septum- gross DNS to left, middle meatus- mucoid secretions on both sides, tongue- glossoptosis, larynx- both vocal cords mobile, Floppy epiglottis (trap door) tall arytenoids, impression- moderate Laryngomalacia.

His 2DECHO showed small PFO-1.4mm, PDA-1.2mm, trivial TR, no PAH.

3. Discussion

Pierre robin sequence or complex is a name given to a birth condition that involves the lower jaw being small in size (micrognathia) or set back from upper jaw (retrognathia). As a result the tongue tends to be displaced back towards throat where it can fall back and obstruct airway (glossoptosis). Most infants have cleft palate involving soft palate leading to u shaped palate. The incidence is greater in girls because the palate takes one week longer time to fuse in girls. Differential diagnosis are Sticklers syndrome, Digeorge syndrome, Treacher Collins syndrome, Fetal Alcohol Syndrome and Velocardiofacial syndrome [2].

In our case the child had micrognathia retrognathia, cleft palate moderate larynogomalacia and stridor . We managed the child conservatively by mechanical ventilation for 3 days SIMV mode for 1 day and later on CPAP and HFNC, anti epileptics were given for seizures, IV fluids and supportive treatment was given. The main modality of treatment is Tracheostomy for airway obstruction and surgical intervention for cleft palate.

4. Conclusion

The case presented to us has micrognathia, retrognathia and cleft palate and on larnygoscopy showed moderate laryngomalacia and it is fitting into pierre robin sequence and the mortality rate in this condition is high 30% [1].

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